

## Early symptoms of Angelman syndrome may include

- Feeding problems.
- Delayed milestones.
- Unusual movements including fine tremors and jerky limb movements.
- Epilepsy.
- Lack of speech development and baby babble.
- Poor sleep patterns.
- Happy demeanour.
- Hand flapping particularly when excited.
- Low muscle tone.

As the children grow they may show:

- A flattened back of head.
- Light skin and hair colour (compared to other family members).
- A tendency to chew objects.
- Severe learning and physical difficulties.
- A similar facial appearance to other children with Angelman syndrome
- A tendency to laugh a great deal.

All patients with Angelman syndrome will benefit from occupational therapy, physiotherapy and speech and language therapy.

Recently the diagnosis of Angelman syndrome is being made more frequently in adults with disabilities. This is because doctors are getting better at recognising it and diagnostic techniques have improved. A person with Angelman syndrome has a normal life expectancy.

## Positive aspects

Most people with Angelman syndrome are sociable, loving and generally happy.

They tend to love water, noisy toys, balloons, balls, TV and music.

Some show considerable understanding of verbal and non-verbal communication.

Some learn sign language and use communication devices.

Sleeping improves with age.

Seizures improve with age.

Some children develop basic speech.

Significant progress can be made by early intervention.

Learning continues throughout life.

Medical research may improve therapies in years to come.

## Translation and interpretation service

Do you have difficulty speaking or understanding English ?

আপনি কি ইংরেজীতে বুঝতে কিংবা বুঝাতে পেরেছেন ? (BENGALI)

क्या आपको अंग्रेजी बोलने या समझने में कठिनाई है ? (HINDI)

તમે ભાષા કાંઈક વાતચીત કરવામાં મુશ્કેલી આવે છે ? (GUJARATI)

ਵਿ ਜੁਹਾਨੂੰ ਅੰਗ੍ਰੇਜ਼ੀ ਬੋਲਣ ਜਾਂ ਸਮਝਣ ਵਿਚ ਦਿੱਕਤ ਹੈ ? (PUNJABI)

Miyey ku adagtahay inaad ku hadasho Ingriisida aad sahamto (SOMALI)

هل لديك مشاكل في فهم اوالتكلم باللغة الانجليزية ؟ (ARABIC)

你有困難講英語或明白英語嗎？ (CANTONESE)

کیا آپ کو انگریزی سمجھنے اور سمجھانے میں دقت پیش آتی ہے؟ (URDU)

☎ Telephone Number 0161 276 6202/6342

**Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).**

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote this leaflet.

Seen in clinic by .....(doctor)

and .....(genetic counsellor)

Telephone number .....

Family reference number .....

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General information

# Angelman Syndrome



INVESTOR IN PEOPLE



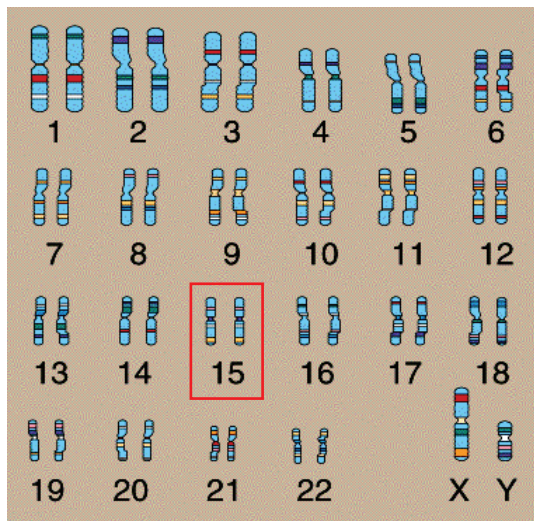
## What is Angelman Syndrome?

Angelman Syndrome is a rare condition that affects 1 person in every 20,000. It is a genetic condition (i.e. it is caused by changes in our genes) which affects parts of the nervous system, particularly the brain. The condition affects both boys and girls.

## What are genes and chromosomes?

Our genes are the unique set of instructions inside every cell of our body. Genes determine our personal characteristics such as eye colour and hair colour. There are many thousands of genes, each carrying a different instruction. As well as determining how we look, our genes control the way each cell of the body works. Specific genes control specific cells.

Chromosomes are made up of thousands of genes. These chromosomes are arranged in 23 pairs, according to size as shown in the picture below. We normally inherit one chromosome from each pair from our mother and one from our father.



There is a support group that provides information packs and has a parent helpline.

A.S.S.E.R.T.  
PO Box 505  
Sittingbourne  
Kent  
ME10 1BR  
Parent Helpline: 01980 652617  
Registered Charity Number: 1021882

## Smoking

Smoking is only permitted within the external designated areas. You may be putting others at risk if you smoke in non-designated areas. The Manchester Stop Smoking Service can be contacted on  
Tel: (0161) 205 5998 ([www.stopsmokingmanchester.co.uk](http://www.stopsmokingmanchester.co.uk)).

## Suggestions, concerns and complaints

If you wish to make a comment, have a concern or want to complain, it is best in the first instance to speak to the manager of the ward or department involved.

The Trust has a Patient Advice and Liaison Service (PALS) who can be contacted on (0161) 276 8686 and via e-mail: [pals@cmmc.nhs.uk](mailto:pals@cmmc.nhs.uk). They will help you if you have a concern, want advice or wish to make a comment or complaint.

Information leaflets about the service are readily available throughout the Trust. Please ask any member of staff for a copy.

## Is it possible to have a second child with Angelman Syndrome?

If you have a child with Angelman syndrome, in most cases, the risk of having a second affected child is low (about 1%).

However, the risk may be higher than this in a few families. The risk depends on the type of genetic alteration your child has and your consultant geneticist will be able to tell you the risk for your family once this has been established.

## For more information

[If you need more advice about any aspect of Angelman Syndrome, you are welcome to contact:](#)

**Genetic Medicine**  
Sixth Floor  
Saint Mary's Hospital  
Oxford Road  
Manchester, M13 9WL  
Telephone: 0161 276 6506 Reception  
Facsimile: 0161 276 6145

Department staffed  
Monday - Friday 9.00 am to 5.00 pm

## Why does Angelman Syndrome occur?

Angelman syndrome is caused by a fault in one of the genes on chromosome 15. There are several different ways in which this gene can be affected including:

- 1) A missing piece of genetic material (deletion) on one of the chromosome 15s. This is the most common cause of Angelman syndrome (75% of cases).
- 2) An alteration (similar to a spelling mistake) in the Angelman gene (10% of cases).
- 3) Both copies of chromosome 15 are inherited from one's father (3% of cases). We usually inherit one chromosome 15 from our mother and one from our father.
- 4) Inheriting a 'switched off' copy of the Angelman gene from one's mother (2%).
- 5) A chromosome rearrangement.

In 10% of cases we are unable to find a genetic cause for Angelman syndrome.